ECFS Diagnostic Network Working Group

(Coordinator: Nico Derichs, Berlin, Germany)

12th Annual Meeting, Warsaw, Poland, 12 - 14th February 2015

Organizing Committee:

Dorota Sands – President of Polish Cystic Fibrosis Society

Monika Mielus – Assistant - Institute of Mother and Child, Warsaw, Poland

Nico Derichs – DNWG Coordinator

Sheila Scheinert – DNWG Assistant

Meeting place:

IBIS Warszawa Stare Miasto (Old Town)
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00-209 WARSAW
POLAND

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Final Program

(Status: 3 Feb 2015)

Thursday 12.02.2015 (14:30-18:00)

13.00 *Lunch*

14.30 Come together

15.00 Welcome – Head of the Institute of Mother and Child - Dr T. Maciejewski, prof. D. Sands

Overview about CF in Poland – Scientific Director of the Institute of Mother and Child –

prof. A. Fijałkowska, prof. D. Sands –President of the Polish Cystic Fibrosis Society

Session 1: CFTR 2015 (Chair: K. De Boeck)

15.30 CFTR models - Piotr Zielenkiewicz (Warsaw, Poland)

16.00 CFTR modulators - state of the art - Aleksander Edelman (Paris, France)

16.30 *- Coffee break -*

16.45 Young Investigator 1:
Global Adaption to CFTR misfolding in Cystic Fibrosis Bronchial Epithelial cells -
Robert Rauscher (Potsdam, Germany)

17.00 Diagnostic implications of "silent" *CFTR* mutations - Zoya Ignatova (Potsdam/Hamburg, Germany)

17.30 Is Primary Sclerosing Cholangitis (PSC) a CFTR-related disorder? Electrophysiological testing and full sequencing of the *CFTR* gene – Michael Wilschanski (Jerusalem, Israel)

Friday 13.02.2015 (9:00-17:00)

Session 2: "Graduating from CFSPID": CF Newborn Screening challenges (Chair: K. Southern, D. Sands)

9.00 Introduction and overview of Cystic Fibrosis Screen Positive Inconclusive Diagnosis (CFSPID) evaluation and management - Kevin Southern (Liverpool, UK)

9.30 CF NBS: IRT step - Mariusz Ołtarzewski (Warsaw, Poland)

10.00 CF NBS: *CFTR* mutation interpretation challenges – Caroline Raynal (Montpellier, France)

10.30 Young Investigator 2: Bilateral sweat test results after CF NBS in Poland – Joanna Jaworska (Warsaw, Poland)

10.45 Young Investigator 3: Genotypes of infants with inconclusive diagnosis after Polish CF NBS – Justyna Milczewska (Warsaw, Poland)

11.00 Challenges associated with the utilisation of next generation sequencing in CF diagnostics: how to assign disease liability of variants detected within the CFTR gene, and beyond - Milan Macek (Prague, Czech Republic)

11.20 - Coffee break -

11.30 Sweat testing in the era of newborn screening - Jürg Barben (St. Gallen, Switzerland)

11.50 Fecal elastase - noninvasive marker of pancreatic function - Jarosław Walkowiak (Poznan, Poland)

12.10 Impulse oscillometry in the evaluation of CF - Waldemar Tomalak (Rabka-Zdroj, Poland)

12.30 Newborn screening for cystic fibrosis: rationale for P.ARG117HIS (R117H) removal from the CFTR mutation panel in France - Anne Munck (Paris, France)

12.45 Discussion Forum: How to optimize country-specific CF NBS programs?

13.00 - Lunch break –

Session 3: Standardising CFTR Biomarker for CF Diagnosis and Clinical Outcome (Chair: N. Derichs)

14.00 Development of an ECFS sweat test guideline: Overview of current practice in Europe – Natalia Cirilli (Ancona, Italy)

14.30 Optimizing sweat test frequency in clinical trials – Francois Vermeulen (Leuven, Belgium)

15.00 ECFS NPD SOP multicenter validation study: Results and diagnostic recommendations – Isabelle Sermet-Gaudelus (Paris, France)

15.30 - Coffee break -

16.00 Young Investigator 4: Developing a multicenter protocol for ratiometric evaluation of B adrenergic/cholinergic sweating in human sweat glands - Denise Peserico (Verona, Italy)

16.15 Young Investigator 5: Optical coherence tomography of the labial salivary glands in cystic fibrosis - Jan Nowak (Poznan, Poland)

16.30 CFTR dysfunction in Primary Ciliary Dyskinesia? - Malena Cohen-Cymberknoh (Jerusalem, Israel)

Saturday 14.02.2015 (8:30-11:00)

Session 4: Journey from Genotype to Phenotype (and retour) (Chair: M. Wilschanski)

8.30 CFTR2, compound heterozygous newborns and sweat test – Carlo Castellani (Verona, Italy)

9.00 R75Q = Non CF-causing mutation? – Elke De Wachter (Brussels, Belgium)

9.30 CFTR3: Personalised characterization of rare *CFTR* mutations – Sheila Scheinert (Berlin, Germany)

9.45 - Coffee break -

10.00 "Patients taking CFTR modulators do not have CF any more"

Pro: Hugo De Jonge (Rotterdam, Netherlands)

Con: Kris De Boeck (Leuven, Belgium)

10.45 Conclusion and perspectives – Nico Derichs (Berlin, Germany)

The meeting is kindly supported by:

Polish Cystic Fibrosis Society